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Abnormal Newborn Screening Tests to be Read on Saturdays and Holidays

Summary

Effective Saturday, March 1, 2014, the S.C. Department of Health and Environmental Control (DHEC) lab will be open on Saturdays and holidays to read newborn screening test results. DHEC staff will call, email and fax the physicians of record (or their on-call physician) to alert them to abnormal test results for conditions with a high risk of mortality or imminent morbidity. If the specimen collection form does not list a physician of record, then DHEC and hospital staff will work together to directly contact the family.

Saturday and holiday notifications will be limited to the following rare, life-threatening diagnoses:

Amino Acid Disorders

Maple Syrup Urine Disease (MSUD)
Citrullinemia
Argininosuccinic Aciduria
Tyrosinemia I

Organic Acid Disorders

Propionic Acidemia
Methylmalonic Acidemia
Isovaleric Acidemia
2-methylbutyryl coA Dehydrogenase Deficiency (2-MBCD)
3-methylcrotonyl coA Carboxylase Deficiency (3-MCC)
Beta Ketothiolase Deficiency (SKAT)
Glutaric Aciduria I

Fatty Acid Disorders

Glutaric Aciduria II
Medium Chain Acyl coA Dehydrogenase Deficiency (MCAD)
Long Chain 3-OH Acyl coA Dehydrogenase Deficiency (LCHAD)
Very Long Chain Acyl coA Dehydrogenase Deficiency (VLCAD)
Carnitine Uptake/Transport Defect (CUD)
Carnitine Palmitoyltransferase I Deficiency (CPT I)
Carnitine Palmitoyltransferase II Deficiency (CPT II)

Others

Galactosemia

Biotinidase deficiency

Congenital Hypothyroidism

Congenital Adrenal Hyperplasia

Resources for Additional Information

newbornscreening@dhec.sc.gov

Categories of Health Alert messages:

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| Health Alert | Conveys the highest level of importance; warrants immediate action or attention. |
| Health Advisory | Provides important information for a specific incident or situation; may not require immediate action. |
| Health Update | Provides updated information regarding an incident or situation; unlikely to require immediate action. |
| Info Service | Provides general information that is not necessarily considered to be of an emergent nature. |